



dilated cardiomyopathy with ataxia syndrome

Dilated cardiomyopathy with ataxia (DCMA) syndrome is an inherited condition characterized by heart problems, movement difficulties, and other features affecting multiple body systems.

Beginning in infancy to early childhood, most people with DCMA syndrome develop dilated cardiomyopathy, which is a condition that weakens and enlarges the heart, preventing it from pumping blood efficiently. Some affected individuals also have long QT syndrome, which is a heart condition that causes the cardiac muscle to take longer than usual to recharge between beats. The irregular heartbeats (arrhythmia) can lead to fainting (syncope) or cardiac arrest and sudden death. Rarely, heart problems improve over time; however, in most cases of DCMA syndrome, affected individuals do not survive past childhood due to heart failure. A small percentage of people with DCMA syndrome have no heart problems at all.

By age 2, children with DCMA syndrome have problems with coordination and balance (ataxia). These movement problems can result in delay of motor skills such as standing and walking, but most older children with DCMA syndrome can walk without support.

In addition to heart problems and movement difficulties, most individuals with DCMA syndrome grow slowly before and after birth, which leads to short stature. Additionally, many affected individuals have mild intellectual disability. Many males with DCMA syndrome have genital abnormalities such as undescended testes (cryptorchidism) or the urethra opening on the underside of the penis (hypospadias). Other common features of DCMA syndrome include unusually small red blood cells (microcytic anemia), which can cause pale skin; an abnormal buildup of fats in the liver (hepatic steatosis), which can damage the liver; and the degeneration of nerve cells that carry visual information from the eyes to the brain (optic nerve atrophy), which can lead to vision loss.

DCMA syndrome is associated with increased levels of a substance called 3-methylglutaconic acid in the urine. The amount of acid does not appear to influence the signs and symptoms of the condition. DCMA syndrome is one of a group of metabolic disorders that can be diagnosed by the presence of increased levels of 3-methylglutaconic acid in urine (3-methylglutaconic aciduria). People with DCMA syndrome also have high urine levels of another acid called 3-methylglutaric acid.

Frequency

DCMA syndrome is a very rare disorder. Approximately 30 cases have been identified in the Dariusleut Hutterite population of the Great Plains region of Canada. Only a few affected individuals have been identified outside this population.

Genetic Changes

Mutations in the *DNAJC19* gene cause DCMA syndrome. The *DNAJC19* gene provides instructions for making a protein found in structures called mitochondria, which are the energy-producing centers of cells. While the exact function of the DNAJC19 protein is unclear, it may regulate the transport of other proteins into and out of mitochondria.

The *DNAJC19* gene mutations that cause DCMA syndrome lead to the production of an abnormally shortened protein that likely has impaired function. Researchers speculate that a lack of functional DNAJC19 protein alters the transport of other proteins into and out of the mitochondria. When too many or too few proteins move in and out of the mitochondria, energy production and mitochondrial survival can be reduced. Tissues that have high energy demands, such as the heart and the brain, are especially susceptible to decreases in cellular energy production. It is likely that this loss of cellular energy damages these and other tissues, leading to heart problems, movement difficulties, and other features of DCMA syndrome.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- 3-methylglutaconic aciduria type V
- DCMA
- DCMA syndrome
- DNAJC19 defect
- MGA type V
- MGA5
- MGCA5

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: 3-methylglutaconic aciduria type V
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857776/>

Other Diagnosis and Management Resources

- Ann & Robert H. Lurie Children's Hospital of Chicago: Cardiomyopathy
<https://www.luriechildrens.org/en-us/care-services/conditions-treatments/cardiomyopathy/Pages/index.aspx>
- Baby's First Test
<http://www.babysfirsttest.org/newborn-screening/conditions/3-methylglutaconic-aciduria>
- MedlinePlus Encyclopedia: Dilated Cardiomyopathy
<https://medlineplus.gov/ency/article/000168.htm>
- National Heart, Lung, and Blood Institute: How is Cardiomyopathy Diagnosed?
<https://www.nhlbi.nih.gov/health/health-topics/topics/cm/diagnosis>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Dilated Cardiomyopathy
<https://medlineplus.gov/ency/article/000168.htm>
- Health Topic: Cardiomyopathy
<https://medlineplus.gov/cardiomyopathy.html>

- Health Topic: Movement Disorders
<https://medlineplus.gov/movementdisorders.html>
- Health Topic: Newborn Screening
<https://medlineplus.gov/newbornscreening.html>

Genetic and Rare Diseases Information Center

- DCMA syndrome
<https://rarediseases.info.nih.gov/diseases/12964/dcma-syndrome>

Educational Resources

- Boston Children's Hospital: Undescended Testes (Cryptorchidism) in Children
<http://www.childrenshospital.org/conditions-and-treatments/conditions/u/undescended-testes-cryptorchidism>
- Cardiomyopathy UK: Dilated Cardiomyopathy
<http://www.cardiomyopathy.org/downloads/life-with-dilated-cardiomyopathy.pdf>
- Centers for Disease Control and Prevention: Intellectual Disability
https://www.cdc.gov/ncbddd/actearly/pdf/parents_pdfs/IntellectualDisability.pdf
- Cincinnati Children's Hospital: Dilated Cardiomyopathy
<https://www.cincinnatichildrens.org/service/c/cardiomyopathy/types/dilated-cardiomyopathy>
- Cleveland Clinic: Dilated Cardiomyopathy
<http://my.clevelandclinic.org/health/articles/dilated-cardiomyopathy>
- Disease InfoSearch: 3-methylglutaconic aciduria type V
<http://www.diseaseinfosearch.org/3-methylglutaconic+aciduria+type+V/7607>
- Johns Hopkins Medicine: What is Ataxia?
http://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/movement_disorders/ataxia/conditions/
- MalaCards: 3-methylglutaconic aciduria, type v
http://www.malacards.org/card/3_methylglutaconic_aciduria_type_v_2
- Merck Manual Home Edition for Patients and Caregivers: Dilated Cardiomyopathy
<http://www.merckmanuals.com/home/heart-and-blood-vessel-disorders/cardiomyopathy/dilated-cardiomyopathy>
- Orphanet: Dilated cardiomyopathy with ataxia
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=66634

- Seattle Children's Hospital: Long QT Syndrome
<http://www.seattlechildrens.org/medical-conditions/heart-blood-conditions/long-qt-syndrome/>
- Washington University, St. Louis: Neuromuscular Disease Center
<http://neuromuscular.wustl.edu/msys/cardiac.html#dcma>

Patient Support and Advocacy Resources

- American Association on Intellectual and Developmental Disabilities (AAIDD)
<http://aaidd.org/>
- American Heart Association: Dilated Cardiomyopathy
http://www.heart.org/HEARTORG/Conditions/More/Cardiomyopathy/Dilated-Cardiomyopathy_UCM_444187_Article.jsp
- Children's Cardiomyopathy Foundation
<http://www.childrenscardiomyopathy.org/>
- National Ataxia Foundation
<http://www.ataxia.org/>
- Organic Acidemia Association
<http://www.oaanews.org/>
- Resource List from the University of Kansas Medical Center: Heart/Cardiology Conditions
<http://www.kumc.edu/gec/support/conghart.html>
- The MAGIC Foundation
<https://www.magicfoundation.org/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22dilated+cardiomyopathy+with+ataxia+syndrome%22+OR+%223-methylglutaconic+aciduria+type+V%22+OR+%22DCMA%22+OR+%22DCMA+syndrome%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28dilated+cardiomyopathy%5BTI%5D%29+AND+%28ataxia%5BTI%5D%29+OR+%28dcma%5BTIAB%5D%29+OR+%28dcma+syndrome%5BTIAB%5D%29+NOT+%28Friedreich%29+NOT+%28Friedreich's%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>

OMIM

- 3-METHYLGLUTACONIC ACIDURIA, TYPE V
<http://omim.org/entry/610198>

Sources for This Summary

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